A 15-Year-Old Male with a Long-Standing History of Blurry Vision

Yicheng Chen, MD; Thomas K. Krummenacher, MD

Introduction:

A 15-year-old healthy male with no other past medical history is referred for longstanding blurred vision in his left eye. On exam his visual acuity is 20/20 in his right eye and 20/40 in his left eye. The intraocular pressures, external exam and anterior chamber exam are all normal. Dilated fundus examination of the right eye is also normal, but the left eye has an elevated pigmented lesion with fibrosis, traction and striae through the fovea. There are also a few spots of intraretinal hemorrhage around the lesion [Image 1]. On FA, there is blockage by areas of pigment along with hyperfluorescence of dilated capillary networks [Image 2].

The history and the exam findings are consistent with a diagnosis of combined hamartoma of the retina and retinal pigment epithelium. Due to his 20/40 vision and stability of symptoms, the decision was made to observe the patient at regular intervals.

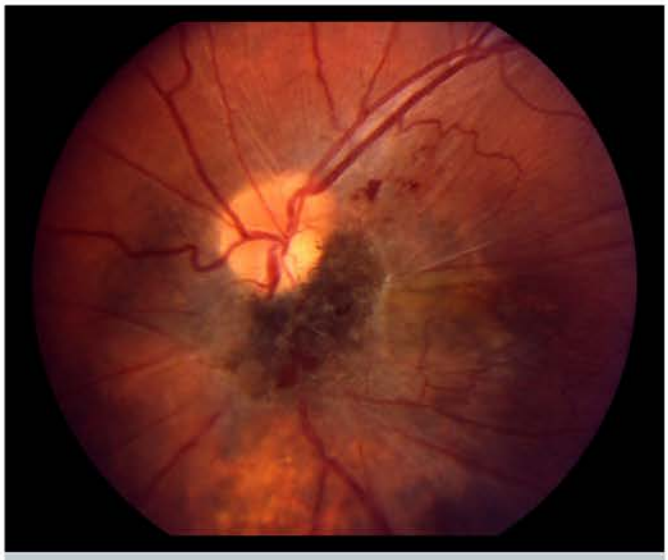

Image 1 - Color photograph of the left eye.

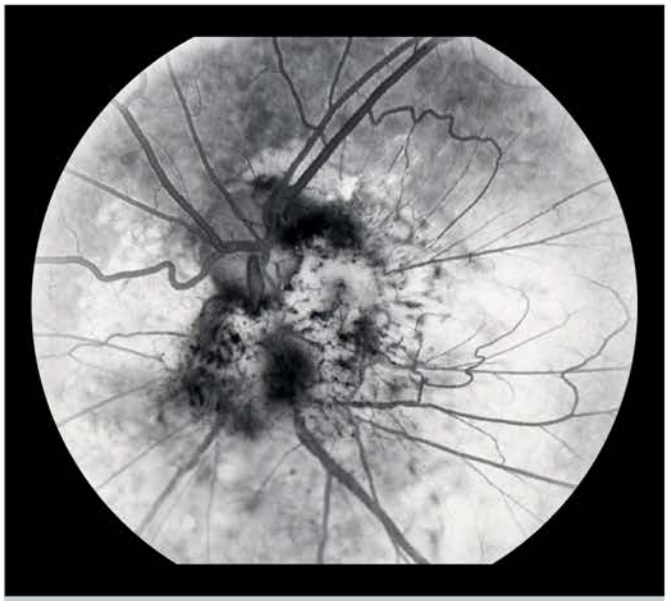

Image 2 - Fluorescein angiography of the left eye.

Discussion:

Combined hamartoma of the retina and retinal pigment epithelium is a benign lesion consisting of glial, vascular and pigment epithelial tissue that can be located in the macula, in the juxtapapillary region or in the periphery. Usually found in children, combined hamartomas of the retina and RPE are thought to be rare congenital lesions. Often characterized by fibrosis, gliosis, epiretinal membrane, traction, and/or fovea dragging, combined hamartomas of the retina and RPE were originally described by Gass[1] as :

(1) Slightly elevated, charcoal grey mass involving the RPE, retina, and overlying vitreous;
(2) Extending in a fanlike projection toward the periphery;
(3) Blending imperceptibly with surrounding RPE;
(4) Covered by thickened grey-white retinal and preretinal tissue;
(5) Showing contraction of the inner surface;

(6) Absence of RPE or choroidal atrophy at the margin; and
(7) With absence of retinal detachment, hemorrhage, exudation, and vitreous inflammation.

Combined hamartomas of the retina and RPE can be associated with a number of systemic conditions, but most strongly with neurofibromatosis type II, especially in bilateral cases. In a large case series, the gender distribution was close to 1:1[2], while another large series found males represented 68% of cases[3]. The median age at diagnosis was 7 months for macular and 8 months for extramacular lesions.

Most common presenting symptoms include vision loss (40%), strabismus (26%), or ocular irritation (5%). A comparison between macular and extramacular cases found that macular epiretinal membrane was associated with 100% of macular cases compared to 15% of extramacular cases, and foveal dragging in 100% vs 55%. Percentage of patients with decreased visual acuity <= 20/200 was 69% vs 25%[3].

Optical coherence tomography and fluorescein angiography can be helpful in establishing the diagnosis. Fluorescein angiography shows early hypofluorescence in areas of hyperpigmentation, while microaneurysms and abnormal dilated capillaries with leakage may be seen in the A/V and later phases[2]. OCT often reveals a distinct epiretinal membrane with secondary retinal folds and striae and adjacent normal retina that gradually thickens into disorganized tissue[4] [Image 3].

Combined hamartomas of the retina and RPE present at young ages, and thus amblyopia treatment is often crucial. Surgical managment of epiretinal membranes is a topic of debate. A small series of 2 cases showed no benefit to visual acuity of surgical removal of the epiretinal membrane[5], but other studies have shown that vision may be improved with surgical intervention[6,7].

Progressive visual loss may occur due to growth of the tumor or complications associated with the tumor such as amblylopia, formation of ERM, retinal holes, retinoschisis, choroidal or retinal neovascularization, retinal heme or retinal detachment. In two large case series, at 4 years the final visual acuity was within two lines of the original visual acuity in 66%, more than two lines worse in 24%, and two or more lines better in 10%. Visual acuity loss of 3 lines or more was seen in 60% of macular involved cases compared to 13% of extramacular cases[2,3].

References:

1) Gass JD. An unusual hamartoma of the pigment epithelium and retina simulating choroidal melanoma and retinoblastoma. Trans Am Ophthalmol Soc. 1973;71:171,83; discussions 184-5.

2) Schachat AP, Shields JA, Fine SL, et al. Combined hamartomas of the retina and retinal pigment epithelium. Ophthalmology. 1984 Dec;91(12):1609-15.

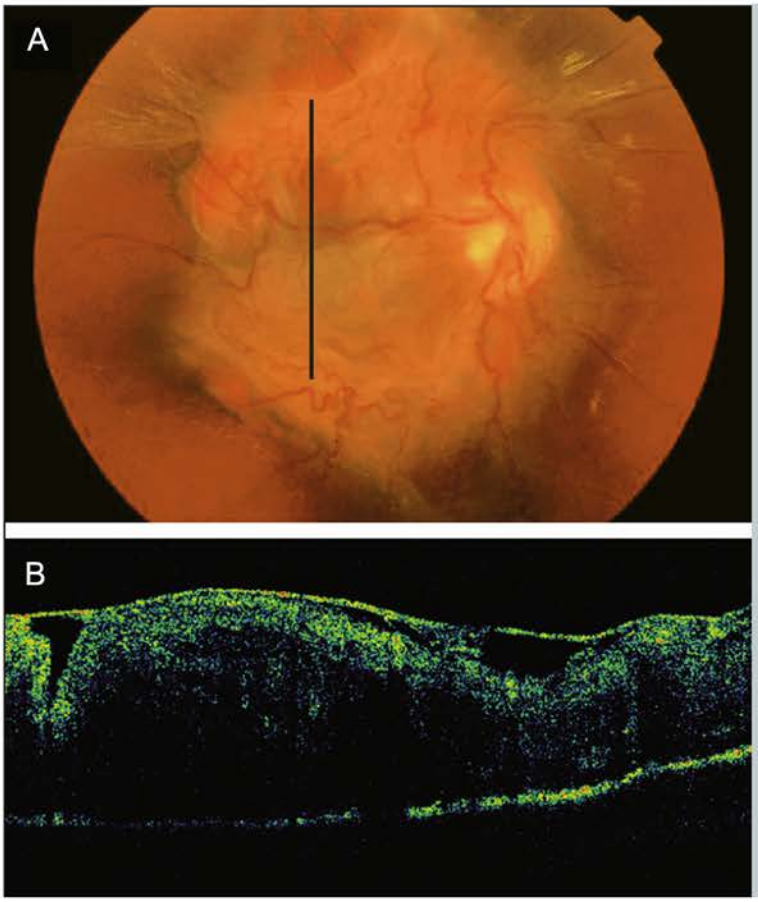

Image 3:
(A) The ill-defined gray retinal mass with retinal vascular traction and retinal thickening is noted.
(B) Vertical optical coherence tomography shows thickened, disorganized retina measuring 1040 µm in thickness. Note the epiretinal membrane inducing undulating folds in the retina.

(Source: Shields CL, Mashayekhi A, Dai VV, Materin MA, Shields JA. Optical coherence tomographic findings of combined hamartoma of the retina and retinal pigment epithelium in 11 patients. Arch Ophthalmol. 2005 Dec; 123(12):1746-50.)

References (cont.):

3) Shields CL, Thangappan A, Hartzell K, Valente P, Pirondini C, Shields JA. Combined hamartoma of the retina and retinal pigment epithelium in 77 consecutive patients visual outcome based on macular versus extra-macular tumor location. Ophthalmology. 2008 Dec;115(12):2246,2252.e3.

4) Shields CL, Mashayekhi A, Dai VV, Materin MA, Shields JA. Optical coherence tomographic findings of combined hamartoma of the retina and retinal pigment epithelium in 11 patients. Arch Ophthalmol. 2005 Dec;123(12):1746-50.

5) McDonald HR, Abrams GW, Burke JM, Neuwirth J. Clinicopathologic results of vitreous surgery for epiretinal membranes in patients with combined retinal and retinal pigment epithelial hamartomas. Am J Ophthalmol. 1985 Dec 15;100(6):806-13.

6) Cohn AD, Quiram PA, Drenser KA, Trese MT, Capone A,Jr. Surgical outcomes of epiretinal membranes associated with combined hamartoma of the retina and retinal pigment epithelium. Retina. 2009 Jun;29(6):825-30.

7) Zhang X, Dong F, Dai R, Yu W. Surgical management of epiretinal membrane in combined hamartomas of the retina and retinal pigment epithelium. Retina. 2010 Feb;30(2):305-9.

UPCOMING EVENTS:

The Retina Institute will soon be opening a new location. . .
Details will be forthcoming!